



ICF Conference
Mayo Clinic - June 21, 2005



Mapping New Vocabularies to the UMLS

Experience with ICF



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Outline

- ◆ Terminology integration
 - *The Unified Medical Language System*
- ◆ Methods
 - Normalizing terms
 - Categorizing terms
 - Recording relations
 - Editing and auditing
- ◆ Experience with ICF



Terminology integration

The Unified Medical Language System

Motivation

- ◆ Started in 1986
- ◆ National Library of Medicine
- ◆ “Long-term R&D project”
- ◆ Complementary to IAIMS

(Integrated Academic
Information Management Systems)

«[...] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

- The first is the variety of ways the same concepts are expressed in different machine-readable sources and by different people.
- The second is the distribution of useful information among many disparate databases and systems.»



Source Vocabularies

(2005AA)

- ◆ 134 source vocabularies
 - 132 contributing concept names
- ◆ ~80 families of vocabularies
 - multiple translations (e.g., MeSH, ICPC, ICD-10)
 - variants (American-English equivalents, Australian extension/adaptation)
 - subsequent editions usually considered distinct families (ICD: 9-10; DSM: IIR-IV)
- ◆ Broad coverage of biomedicine
- ◆ Common presentation



Biomedical terminologies

◆ General vocabularies

- anatomy (UWDA, Neuronames)
- drugs (RxNorm, First DataBank, Micromedex)
- medical devices (UMD, SPN)

◆ Several perspectives

- clinical terms (SNOMED CT)
- information sciences (MeSH, CRISP)
- administrative terminologies (ICD-9-CM, CPT-4)
- data exchange terminologies (HL7, LOINC)



Biomedical terminologies (cont'd)

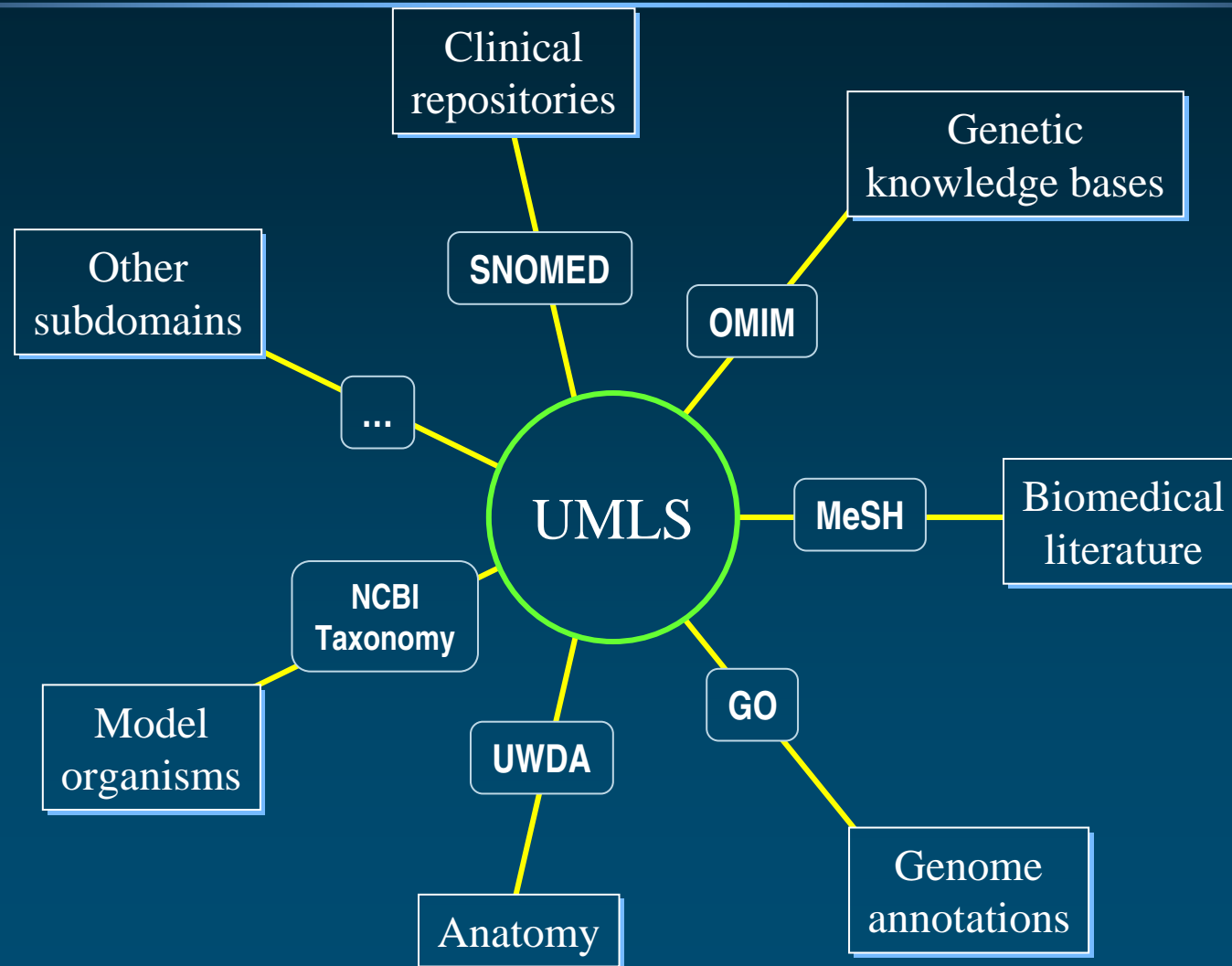
◆ Specialized vocabularies

- nursing (NIC, NOC, NANDA, Omaha, PCDS)
- dentistry (CDT)
- psychiatry (DSM, APA)
- adverse reactions (COSTART, WHO ART)
- primary care (ICPC)
- genomics (GO, OMIM, HUGO)

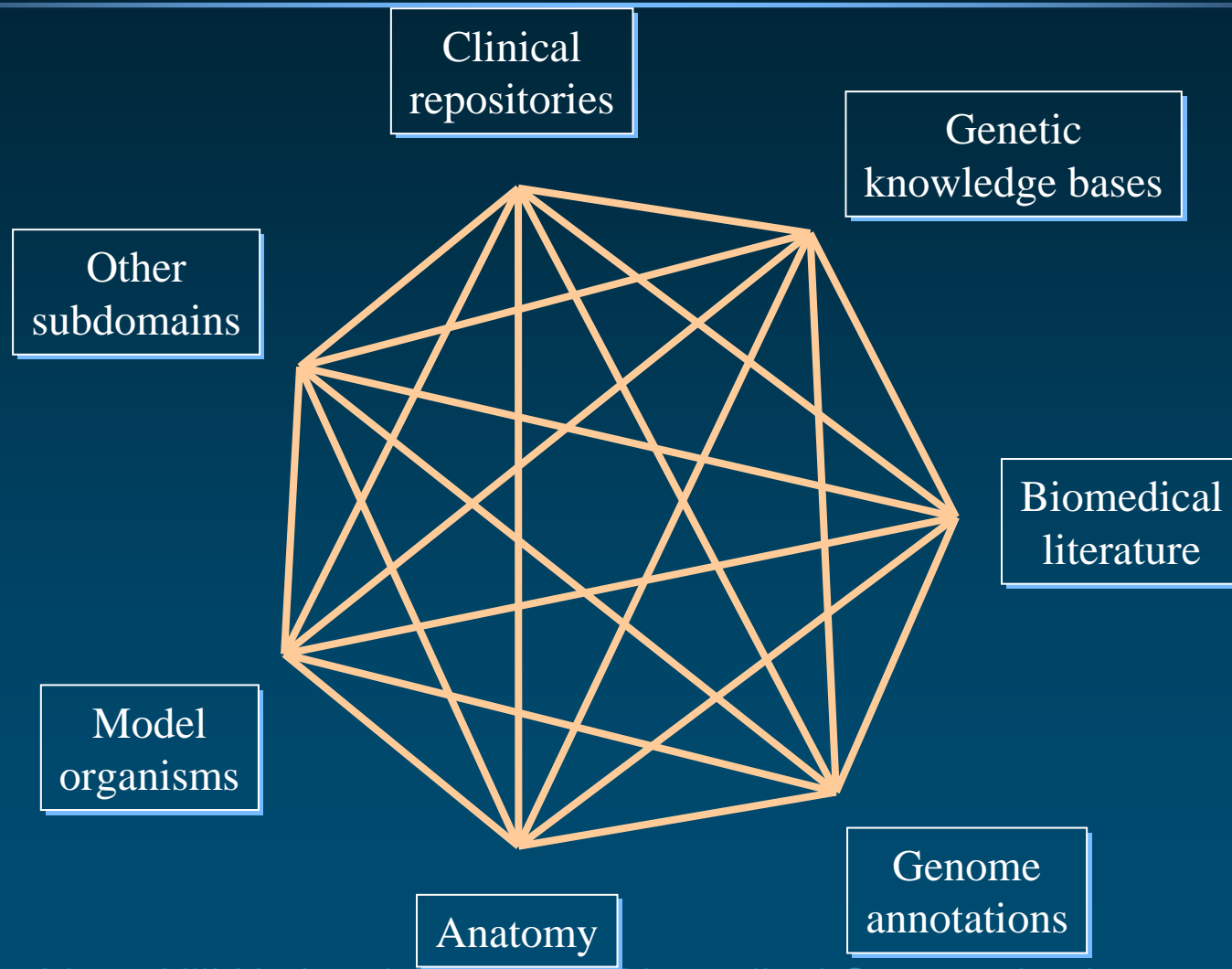
◆ Terminology of knowledge bases (AI/Rheum, DXplain, QMR)

The UMLS serves as a vehicle for the regulatory standards (HIPAA, CHI)

Integrating subdomains



Integrating subdomains



UMLS: 3 components

◆ Metathesaurus

- Concepts
- Inter-concept relationships

◆ Semantic Network

- Semantic types
- Semantic network relationships

◆ Lexical resources

- SPECIALIST Lexicon
- Lexical tools



Addison's Disease in medical vocabularies

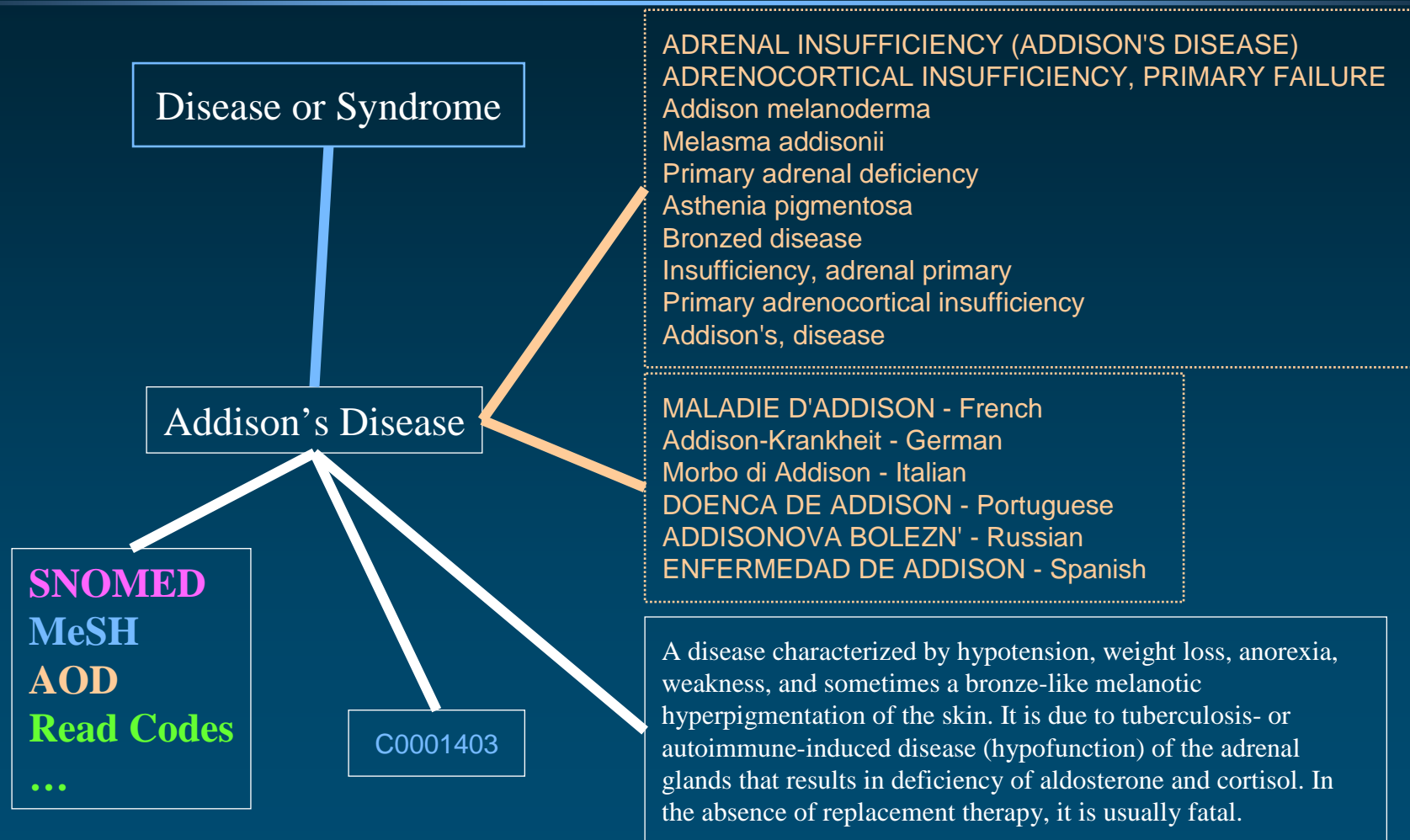
◆ Synonyms: different terms

- | | | |
|--|---|----------------------|
| • Addisonian syndrome | } | eponym |
| • Bronzed disease | | |
| • Addison melanoderma | } | symptoms |
| • Asthenia pigmentosa | | |
| • Primary adrenal deficiency | } | clinical
variants |
| • Primary adrenal insufficiency | | |
| • Primary adrenocortical insufficiency | | |
| • Chronic adrenocortical insufficiency | | |

◆ Contexts: different hierarchies



Addison's Disease: Concept



Metathesaurus Concepts (2005AA)

- ◆ Concept (~ 1.2M) CUI
 - Set of synonymous concept names
- ◆ Term (~ 4.2 M) LUI
 - Set of normalized names
- ◆ String (~ 4.7M) SUI
 - Distinct concept name
- ◆ Atom (~ 5.5M) AUI
 - Concept name in a given source

A0000001 headache (source 1)
A0000002 headache (source 2)
S0000001

A0000003 Headache (source 1)
A0000004 Headache (source 2)
S0000002

L0000001

A0000005 Cephalgia (source 1)
S0000003

L0000002

C0000001



Cluster of synonymous terms

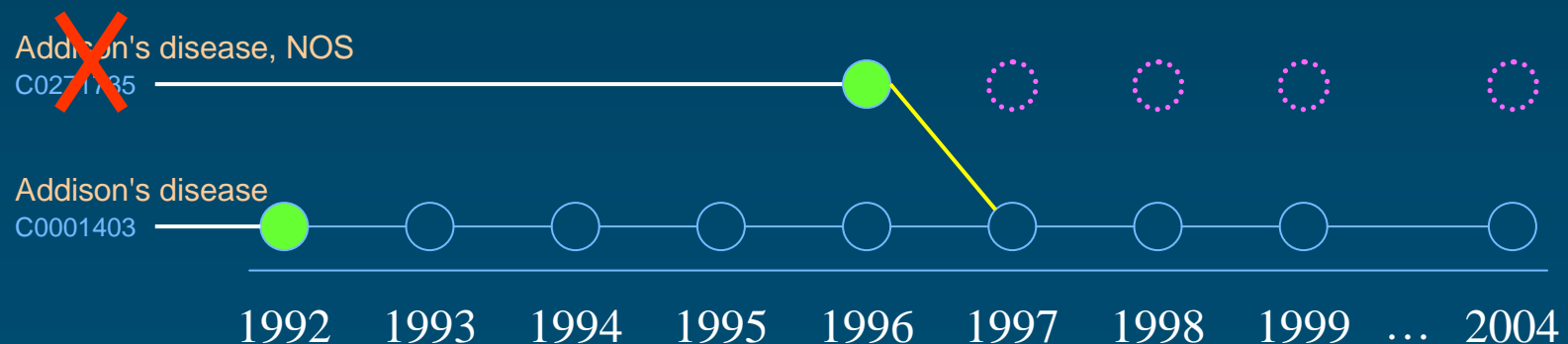
Concept
C0001621

Term L0001621	<p>S0011232 <i>Adrenal Gland Diseases</i></p> <p>S0011231 Adrenal Gland Disease</p> <p>S0000441 Disease of adrenal gland [...]</p> <p>S0481705 Disease of adrenal gland, NOS</p> <p>S0220090 Disease, adrenal gland</p> <p>S0044801 Gland Disease, Adrenal</p>
Term L0041793	<p>S0860744 <i>Disorder of adrenal gland, unspecified</i></p> <p>S0217833 Unspecified disorder of adrenal glands</p>
Term L0161347	<p>S0225481 <i>ADRENAL DISORDER</i></p> <p>S0627685 DISORDER ADRENAL (NOS) [...]</p>
Term L0181041	<p>S0632950 <i>Disorder of adrenal gland</i></p> <p>S0354509 Adrenal Gland Disorders [...]</p>
Term L0368399	<p>S0586222 <i>Adrenal disease</i></p> <p>S0466921 ADRENAL DISEASE, NOS [...]</p>
Term L1279026	<p>S1520972 <i>Nebennierenkrankheiten</i> GER</p>
Term L0162317	<p>S0226798 <i>SURRENALE, MALADIES</i> FRE [...]</p>



Metathesaurus Evolution over time

- ◆ Concepts never die (in principle)
 - CUIs are permanent identifiers
- ◆ What happens when they do die (in reality)?
 - Concepts can merge or split
 - Resulting in new concepts and deletions



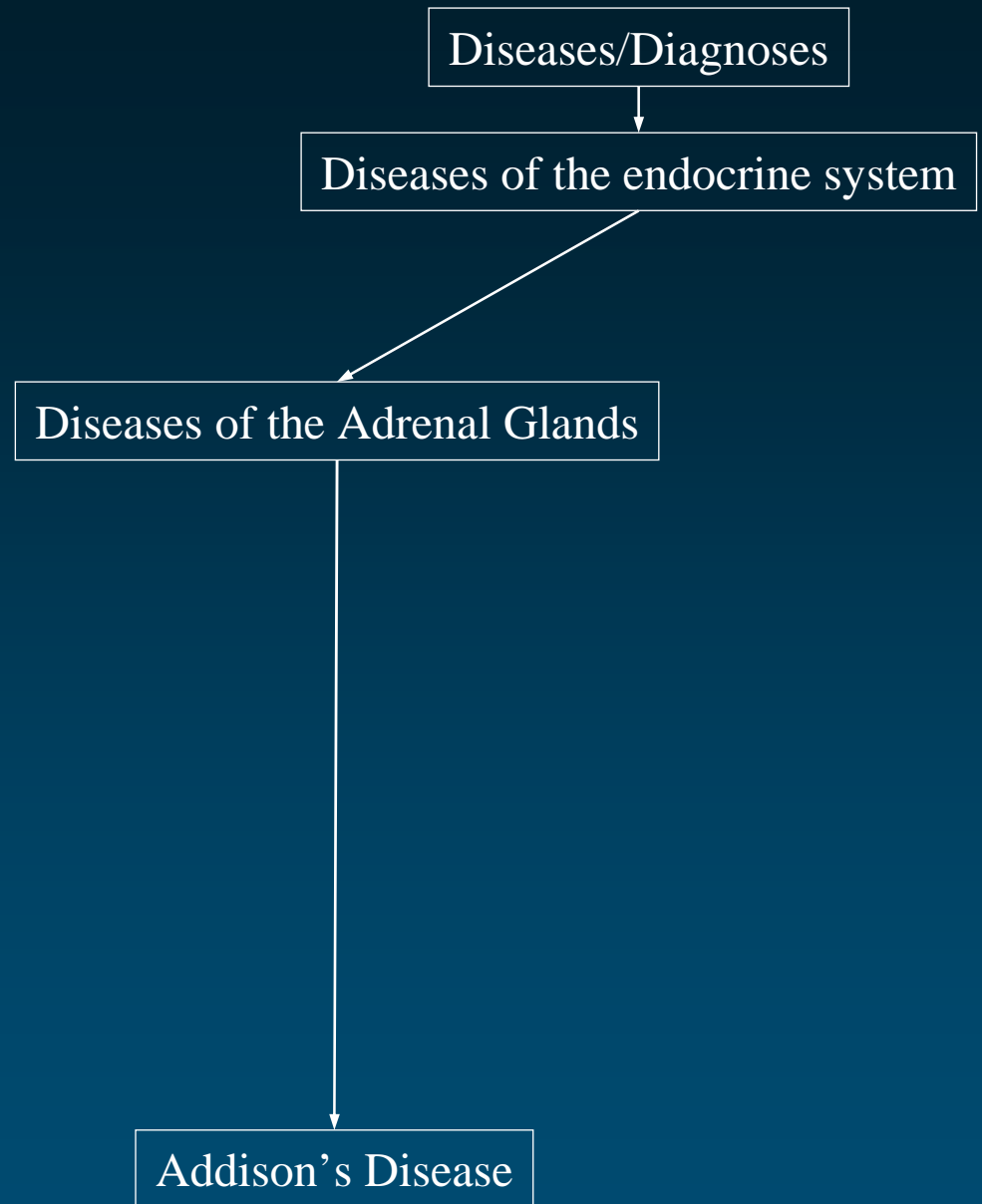
Metathesaurus Relationships

- ◆ Symbolic relations: ~9 M pairs of concepts
 - ◆ Statistical relations : ~7 M pairs of concepts (co-occurring concepts)
 - ◆ Mapping relations: 100,000 pairs of concepts
-

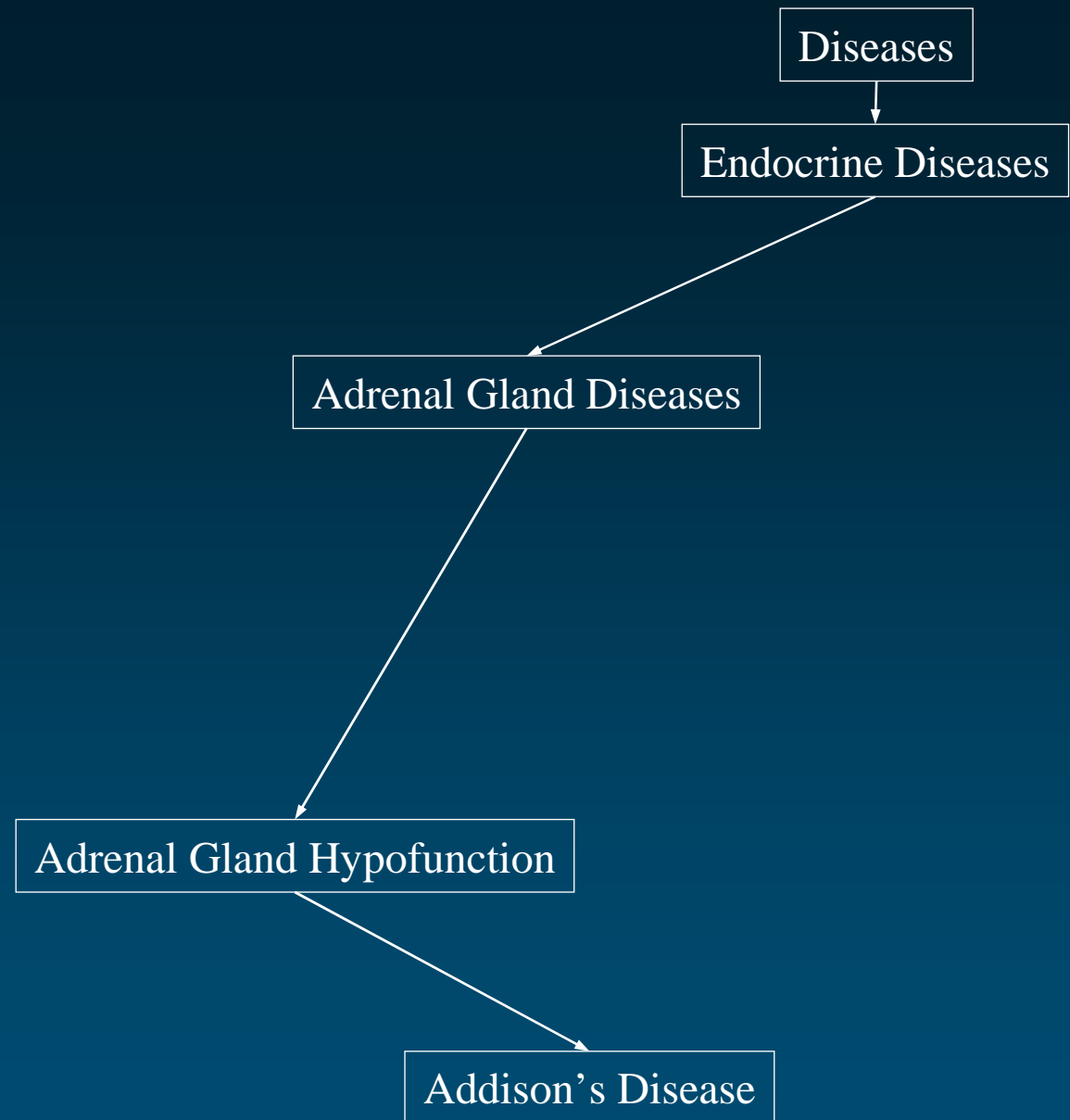
- ◆ Categorization: Relationships between concepts and semantic types from the Semantic Network



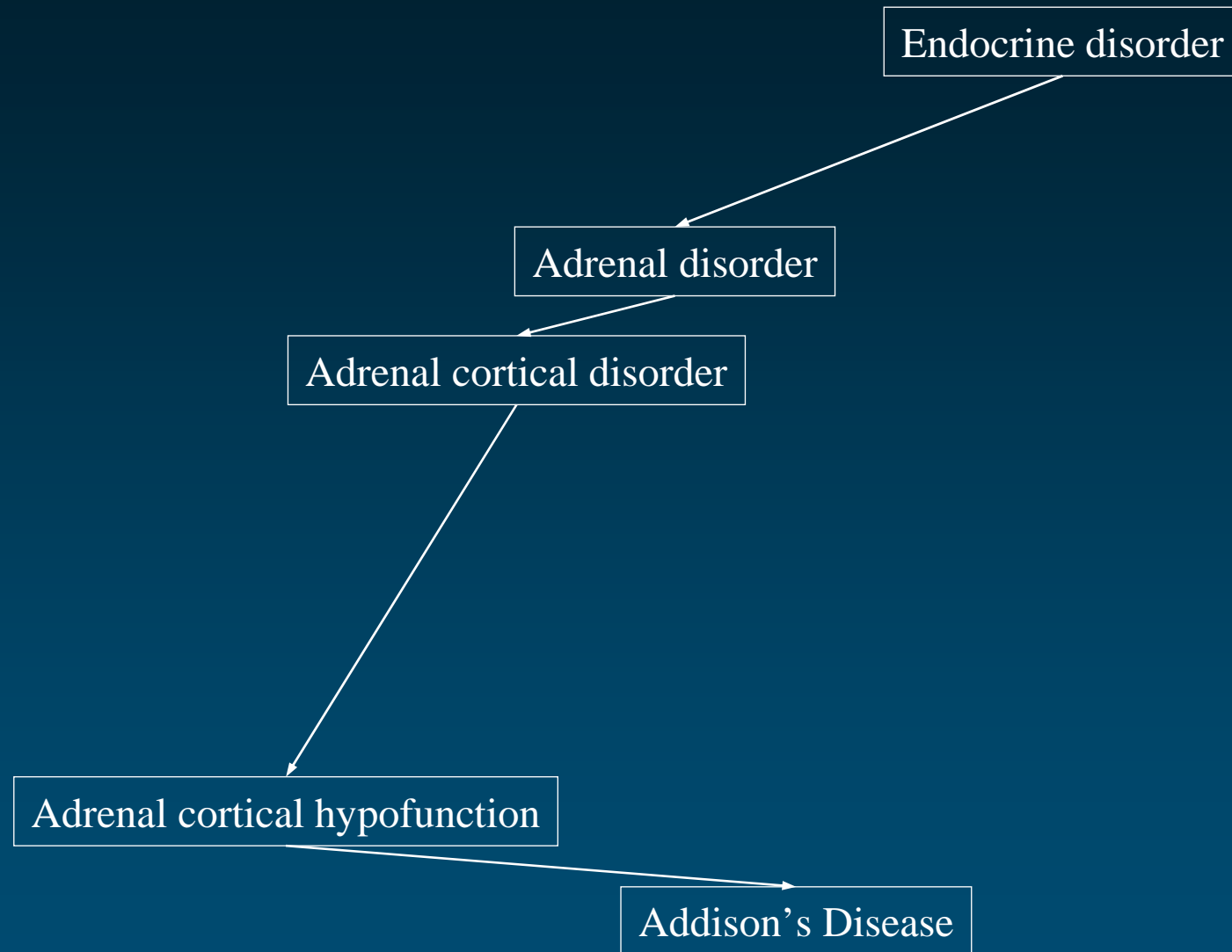
SNOMED International



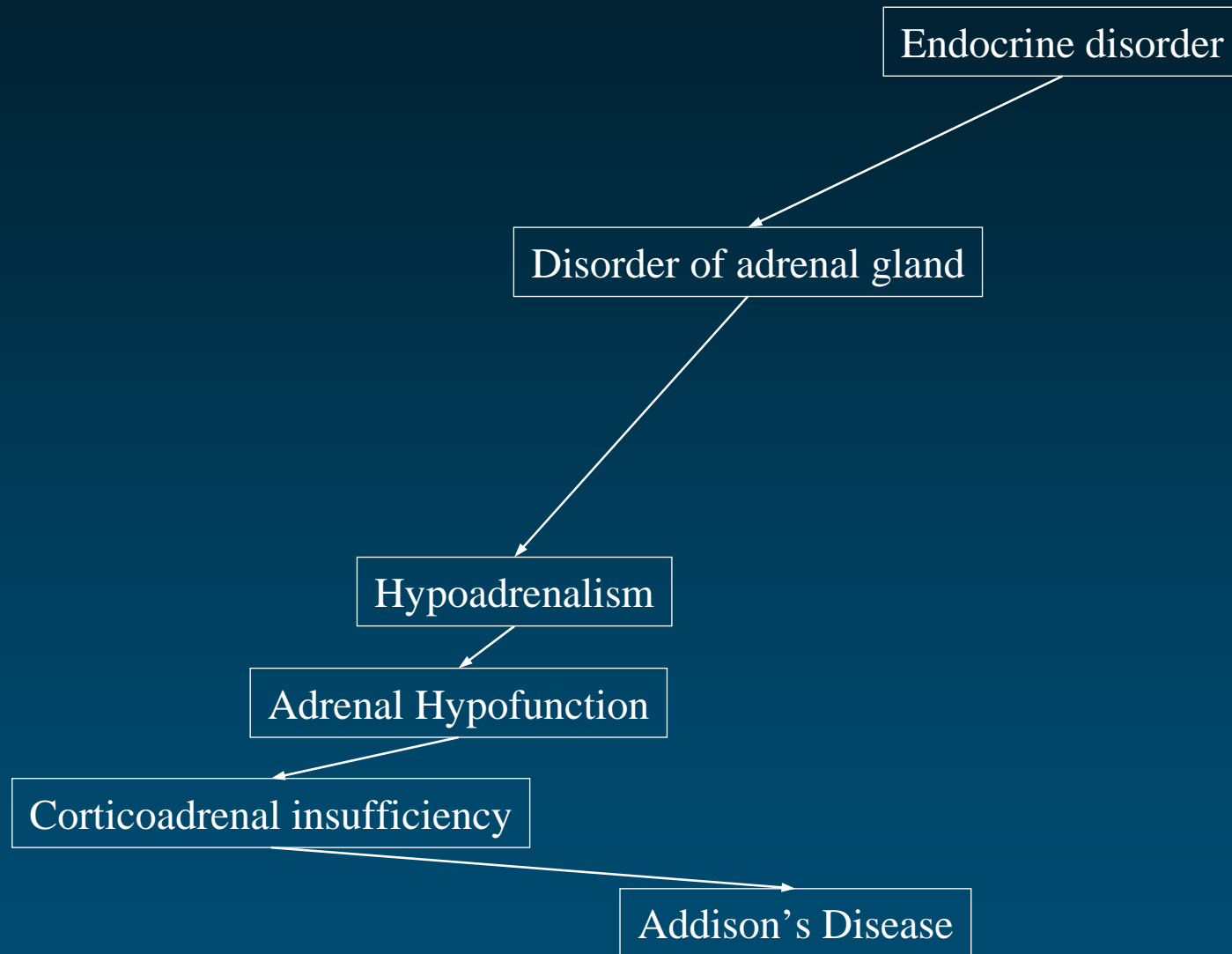
MeSH



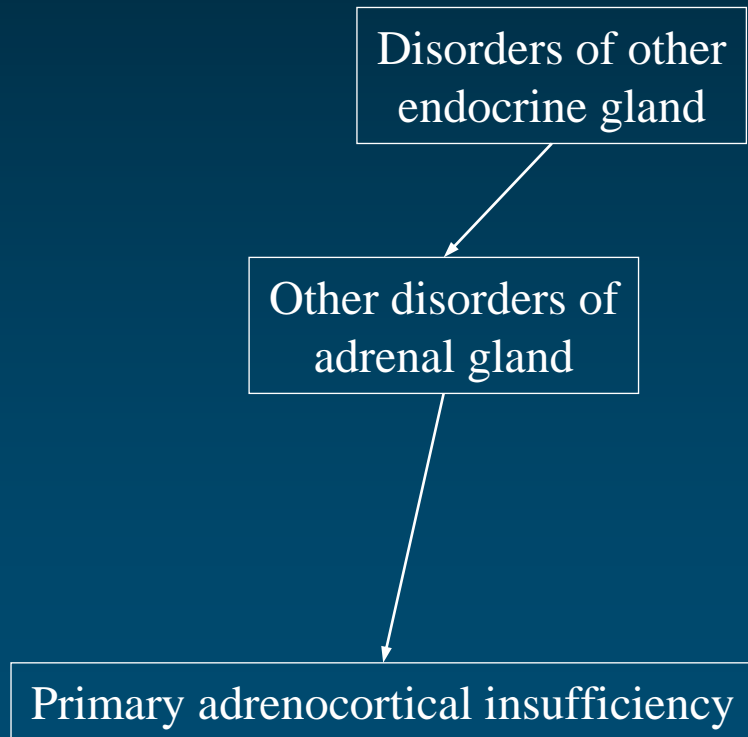
AOD



Read Codes

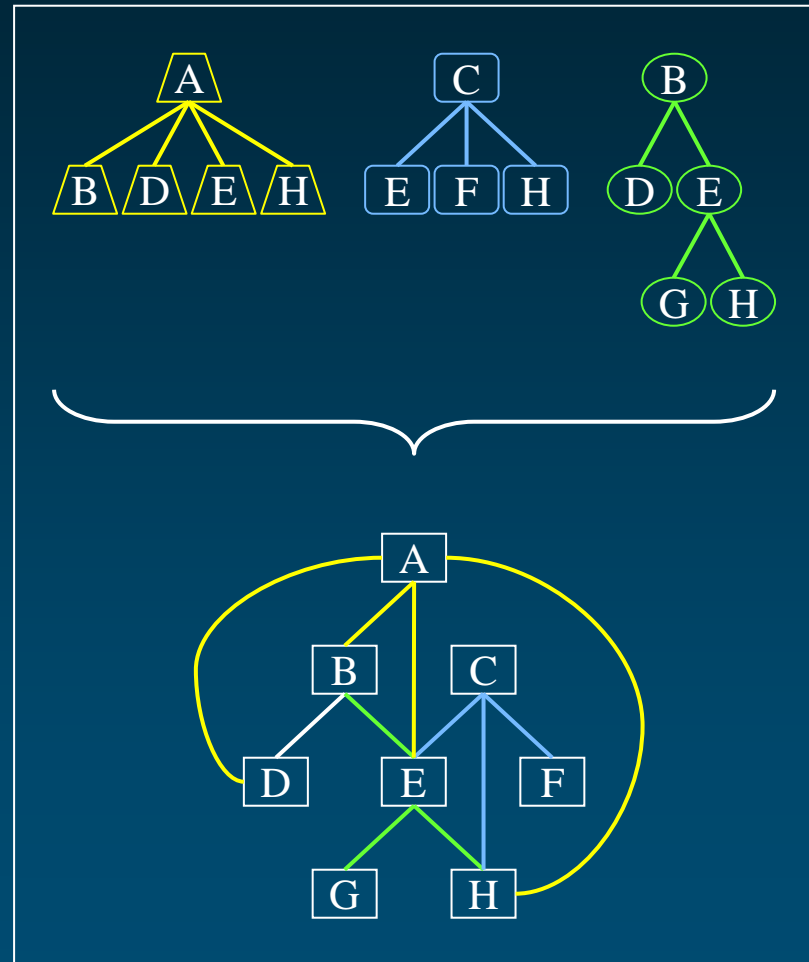


ICD-10



Organize concepts

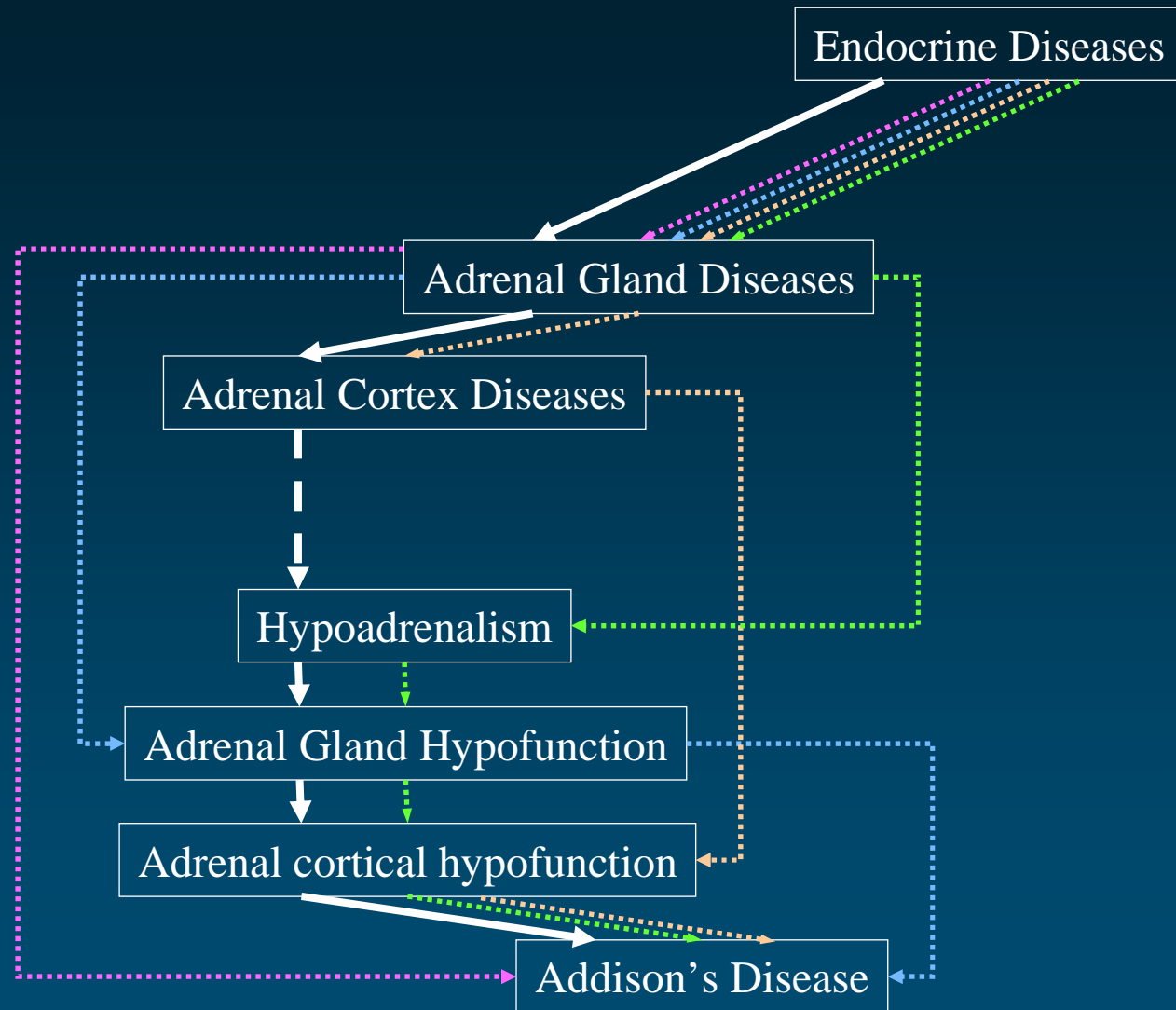
- ◆ Inter-concept relationships: hierarchies from the source vocabularies
- ◆ Redundancy: multiple paths
- ◆ One graph instead of multiple trees (multiple inheritance)

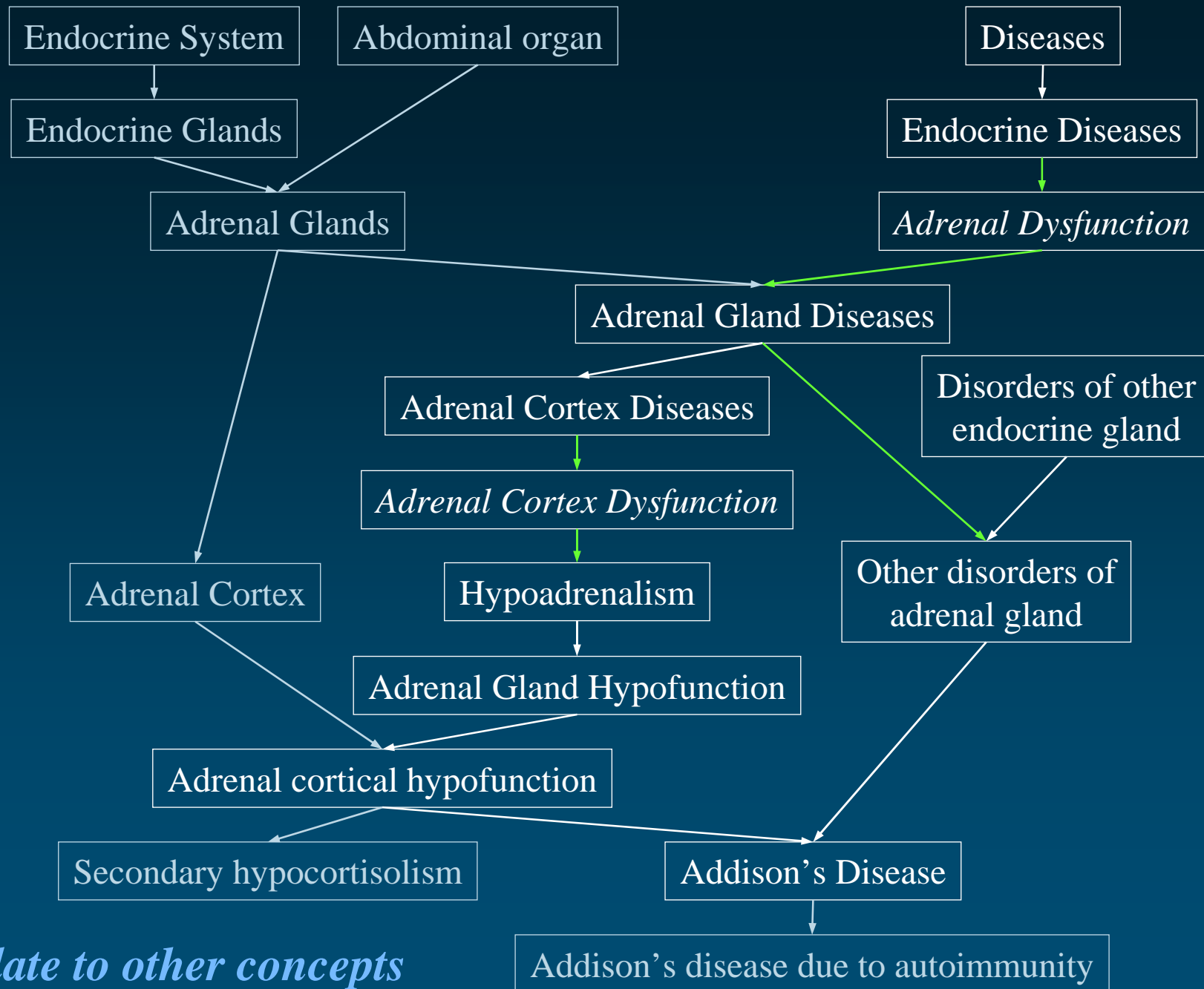


organize concepts

SNOMED
MeSH
AOD
Read Codes

UMLS





Symbolic relations

◆ Relation

- Pair of “atom” identifiers
- Type
- Attribute (if any)
- List of sources (for type and attribute)

◆ Semantics of the relationship: defined by its *type* [and *attribute*]

Source transparency: the information
is recorded at the “atom” level



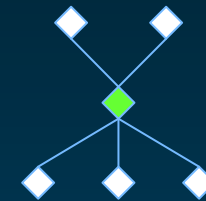
Symbolic relationships Type

◆ Hierarchical

- Parent / Child
- Broader / Narrower than

PAR / CHD

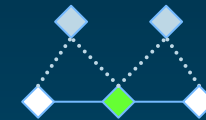
RB / RN



◆ Derived from hierarchies

- Siblings (children of parents)

SIB



◆ Associative

- Other

RO



◆ Various flavors of near-synonymy

- Similar
- Source asserted synonymy
- Possible synonymy

RL

SY

RQ



Symbolic relationships Attribute

◆ Hierarchical

- isa (is-a-kind-of)
- part-of

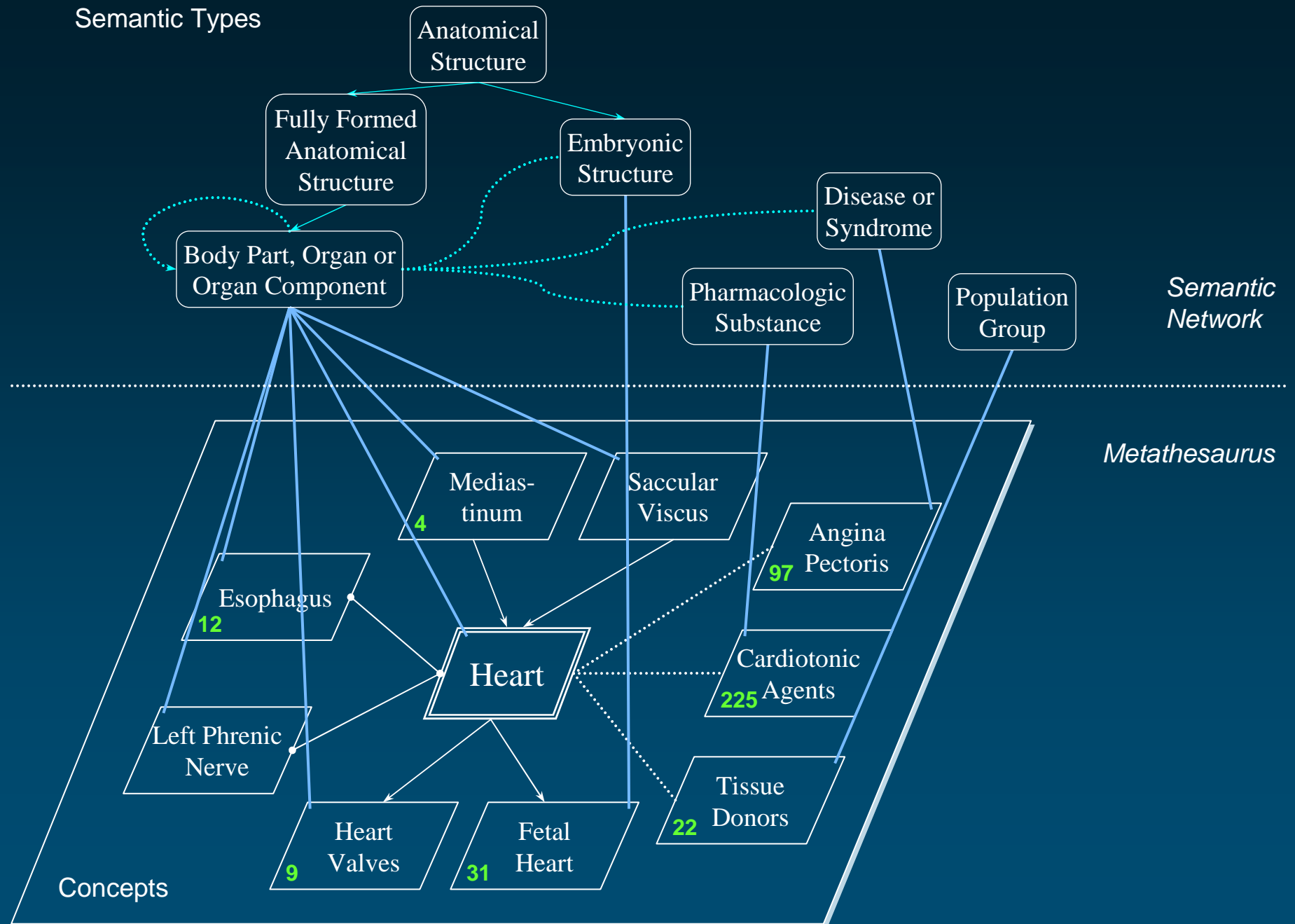
◆ Associative

- location-of
- caused-by
- treats
- ...

◆ Cross-references (mapping)



Semantic Types



Terminology integration methods

How do they do that?

- ◆ Integrating terms
Lexical knowledge
- ◆ Categorizing concepts
Semantic pre-processing
- ◆ Integrating relations
Recording relations
- ◆ Editing and auditing
UMLS editors



Terminology integration methods

Lexical knowledge

Lexical knowledge

Adrenal gland diseases

Adrenal disorder

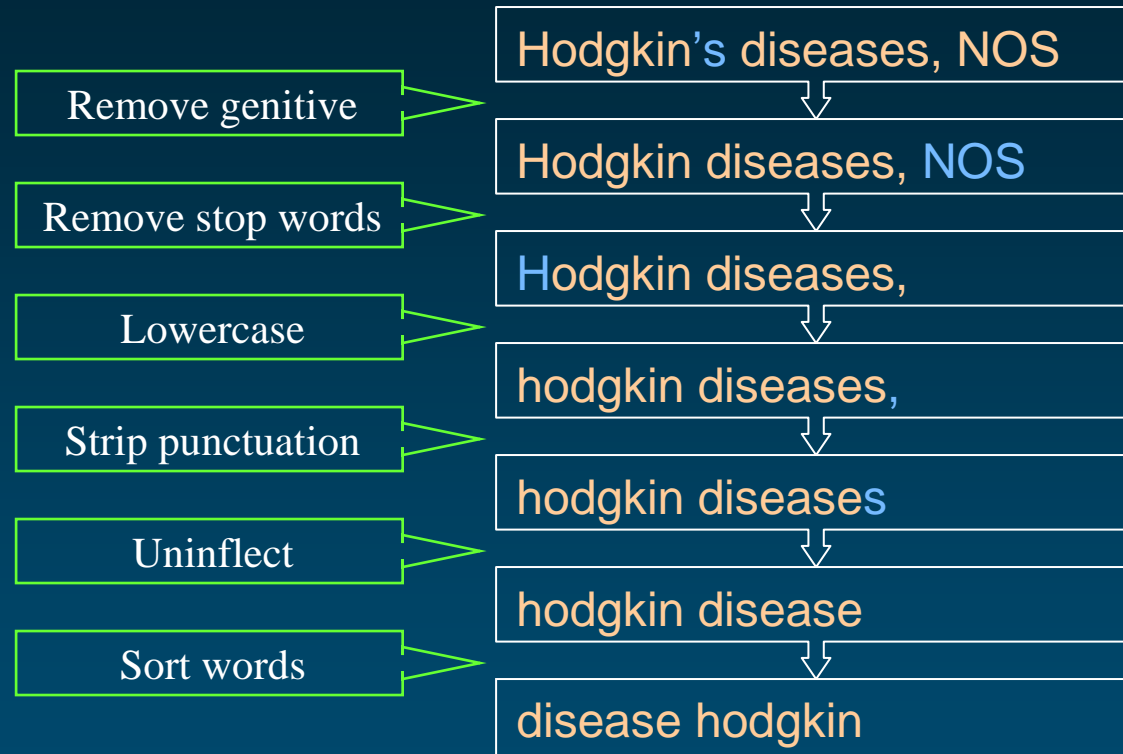
Disorder of adrenal gland

Diseases of the adrenal glands

C0001621



Normalization



Normalization: Example

Hodgkin Disease
HODGKINS DISEASE
Hodgkin's Disease
Disease, Hodgkin's
Hodgkin's, disease
HODGKIN'S DISEASE
Hodgkin's disease
Hodgkins Disease
Hodgkin's disease NOS
Hodgkin's disease, NOS
Disease, Hodgkins
Diseases, Hodgkins
Hodgkins Diseases
Hodgkins disease
hodgkin's disease
Disease, Hodgkin

normalize

disease hodgkin



Lexical tools

- ◆ To manage lexical variation in biomedical terminologies
- ◆ Major tools
 - Normalization
 - Indexes
 - Lexical Variant Generation program (lvg)
- ◆ Based on the SPECIALIST Lexicon
- ◆ Used by noun phrase extractors, search engines





Integrating terms Examples

◆ Exact match

- Original term: Pain in back (b28013)
- Concept mapped to: Back Pain (C0003862)

Pain in back present in the Metathesaurus (from the Read Codes)

◆ Match after normalization

- Original term: Pain in joints (b28016)
- Normalized term: joint pain
- Concept mapped to: Arthralgia (C0003862)

Joint pain is a synonym for Arthralgia





Integrating terms Examples

◆ No match found

- Radiating pain in body part (b2801) *Too general*
- Radiating pain in a dermatome (b2803) *Too specific*
- Pain in stomach or abdomen (b28012) *Coordination*

e215	<u>Population</u>	(→ C0032659)
e2150	Demographic change	(→ C0681668)
e2151	Population density	(→ C0032665)
e2158	<u>Population</u> , other specified	←
e2159	<u>Population</u> , unspecified	



Integrating terms Examples

◆ Multiple matches

- Impulse control (b1304)
 - Impulse control (C0150632)
 - Impulse control training (C0262701)
 - Ability to control impulses (C0517616)

- Frontal lobe (s11000)
 - frontal lobe (C0016733)
 - Entire frontal lobe (C1268977) } *SNOMED CT distinction*

- Bites (b5101)
 - Biting (C0005658)
 - 2-(4-ethoxybenzyl)-1-diethylaminoethyl-5-isothiocyanatobenzimidazole (C0045724)

synonym for BIT alkylating agent



Terminology integration methods

Semantic pre-processing

Semantic pre-processing

- ◆ Metadata in the source vocabularies
- ◆ Tentative categorization
- ◆ Positive (or negative) evidence for tentative synonymy relations based on lexical features



Semantic pre-processing in practice

◆ Mapping between

- Semantic types (UMLS Semantic Network)
- Semantics of a given subset of a terminology

◆ Semantic Network

- 135 semantic types (high-level categories)
- 2 hierarchies for **Entity** and **Event**
- Examples
 - **Disease or Syndrome**
 - **Body Part, Organ, or Organ Component**
 - **Mental Process**



UMLS Semantic Groups

- ◆ ACTI Activities & Behaviors
- ◆ ANAT Anatomy
- ◆ CHEM Chemicals & Drugs
- ◆ CONC Concepts & Ideas
- ◆ DEVI Devices
- ◆ DISO Disorders
- ◆ GENE Genes & Molecular Sequences
- ◆ GEOG Geographic Areas
- ◆ LIVB Living Beings
- ◆ OBJC Objects
- ◆ OCCU Occupations
- ◆ ORGA Organizations
- ◆ PHEN Phenomena
- ◆ PHYS Physiology
- ◆ PROC Procedures

- Acquired Abnormality
- Anatomical Abnormality
- Cell or Molecular Dysfunction
- Congenital Abnormality
- Disease or Syndrome
- Experimental Model of Disease
- Finding
- Injury or Poisoning
- Mental or Behavioral Dysfunction
- Neoplastic Process
- Pathologic Function
- Sign or Symptom



Semantic areas in ICF

- ◆ b BODY FUNCTIONS
 - **Physiology**
 - Sign or Symptom
 - Finding
 - Biologic Function
 - Individual Behavior
- ◆ s BODY STRUCTURES
 - **Anatomy**
- ◆ d ACTIVITIES AND PARTICIPATION
 - **Physiology**
 - **Activities & Behaviors**
 - Machine Activity
 - Sign or Symptom
 - Finding
 - Educational Activity



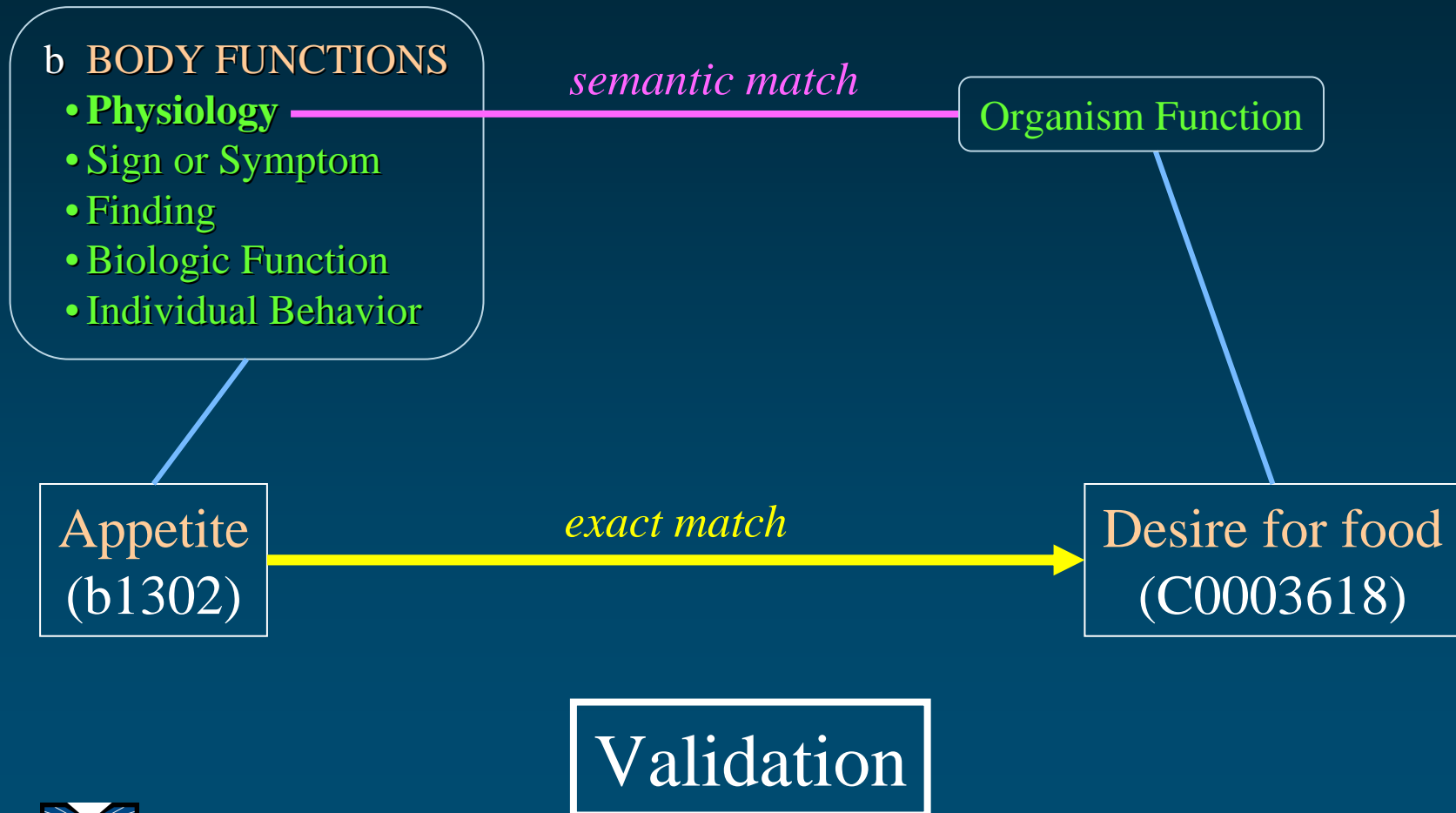
Semantic areas in ICF

- ◆ e1 PRODUCTS AND TECHNOLOGY
 - ????
- ◆ e2 NATURAL ENVIRONMENT AND HUMAN-MADE CHANGES TO ENVIRONMENT
 - **Phenomena**
- ◆ e3 SUPPORT AND RELATIONSHIPS
 - Family Group
 - Population Group
 - Professional or Occupational Group
- ◆ e4 ATTITUDES
 - ????
- ◆ e5 SERVICES, SYSTEMS AND POLICIES
 - Governmental or Regulatory Activity
 - Regulation or Law



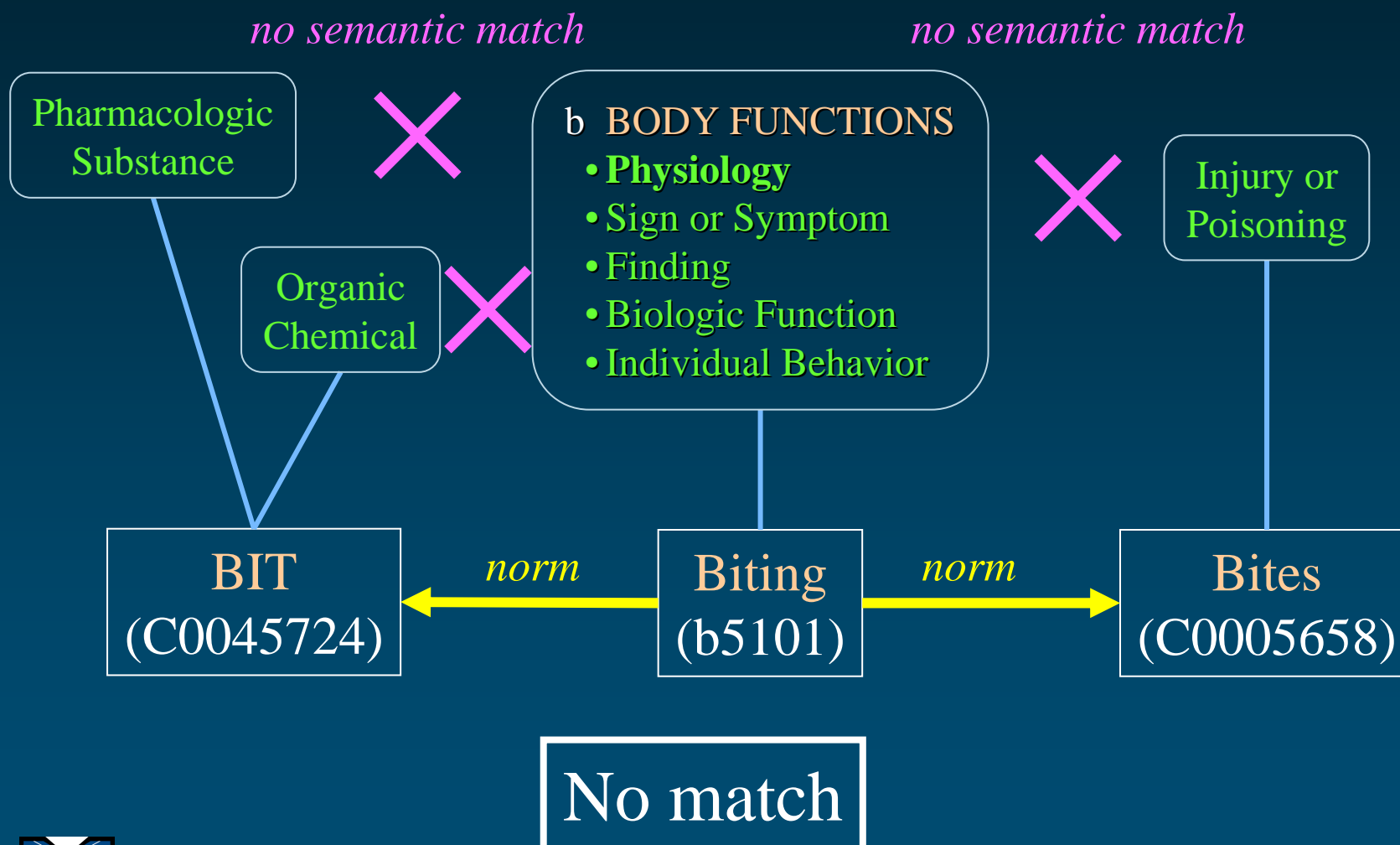


Semantic pre-processing Examples



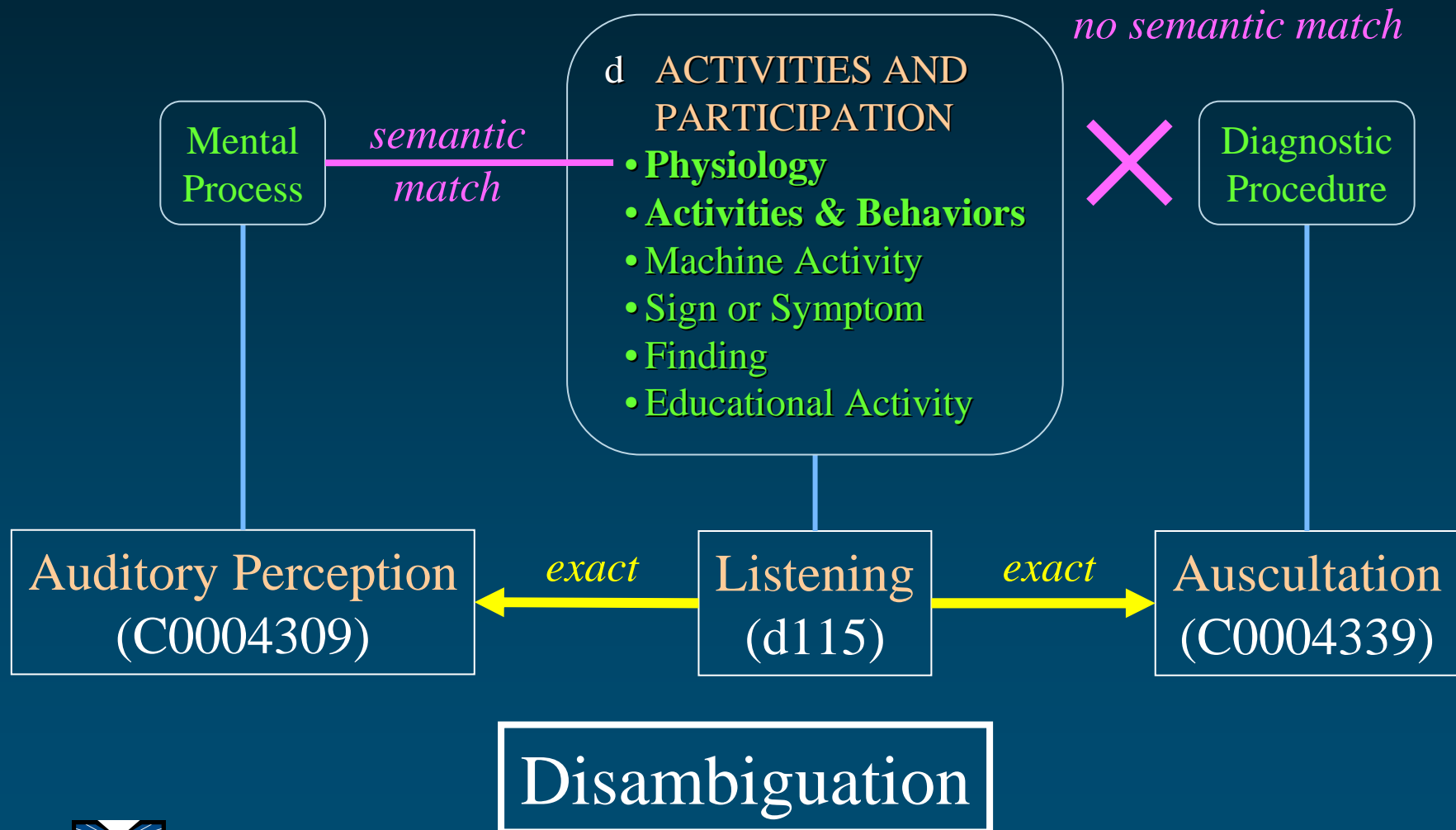


Semantic pre-processing Examples





Semantic pre-processing Examples



Terminology integration methods

Recording relations

Recording relations

- ◆ Relations
 - Recorded at the term (atom) level
 - Aggregated at the concept level
- ◆ Once integrated into the UMLS, ICF relations participate to the Metathesaurus graph
- ◆ Possibly redundant with relations from other sources



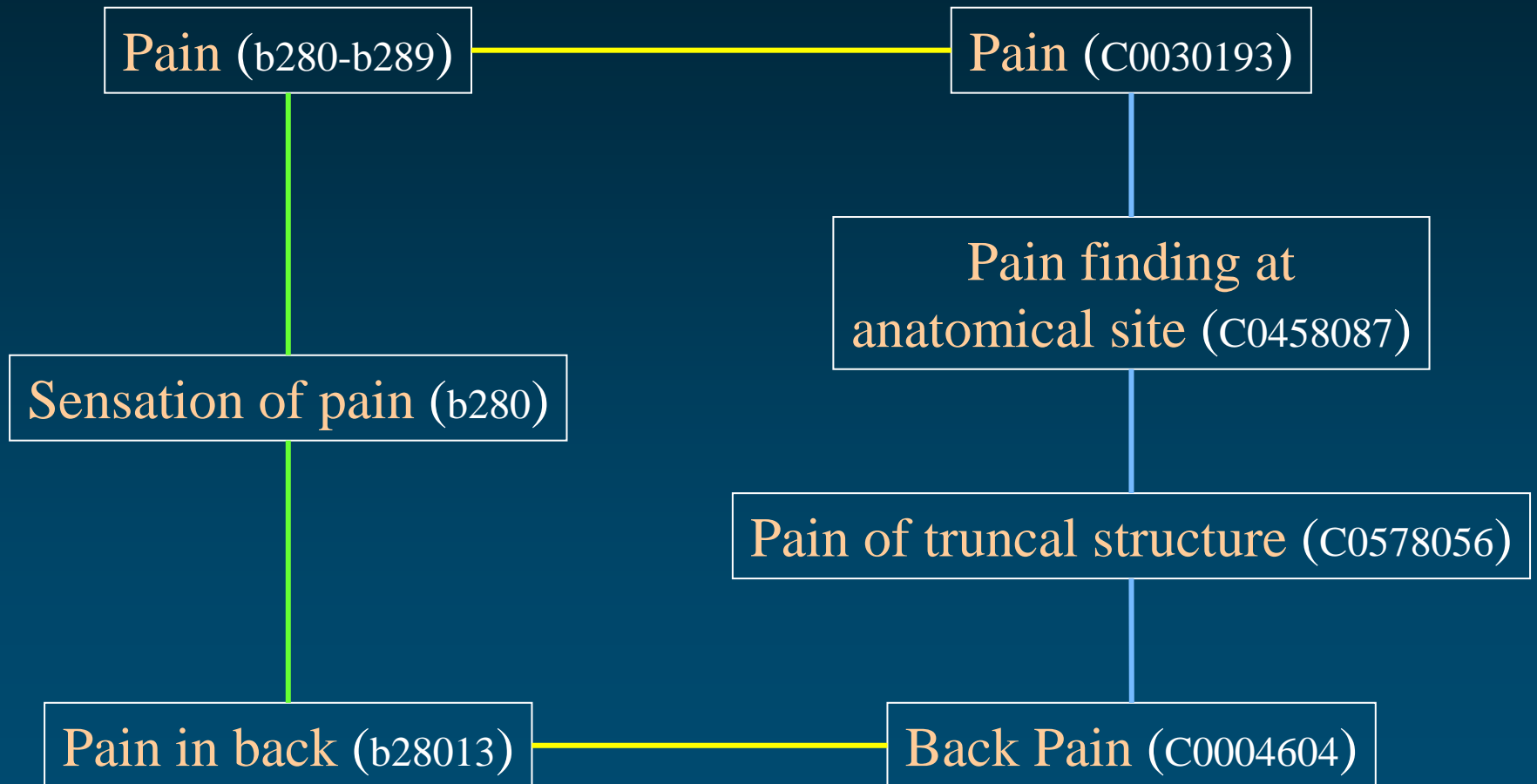


ICF relations in the Metathesaurus

- ◆ ICF hierarchical relations in the UMLS
 - REL: parent/child
 - RELA: none
 - SAB: ICF
- ◆ Other relations?

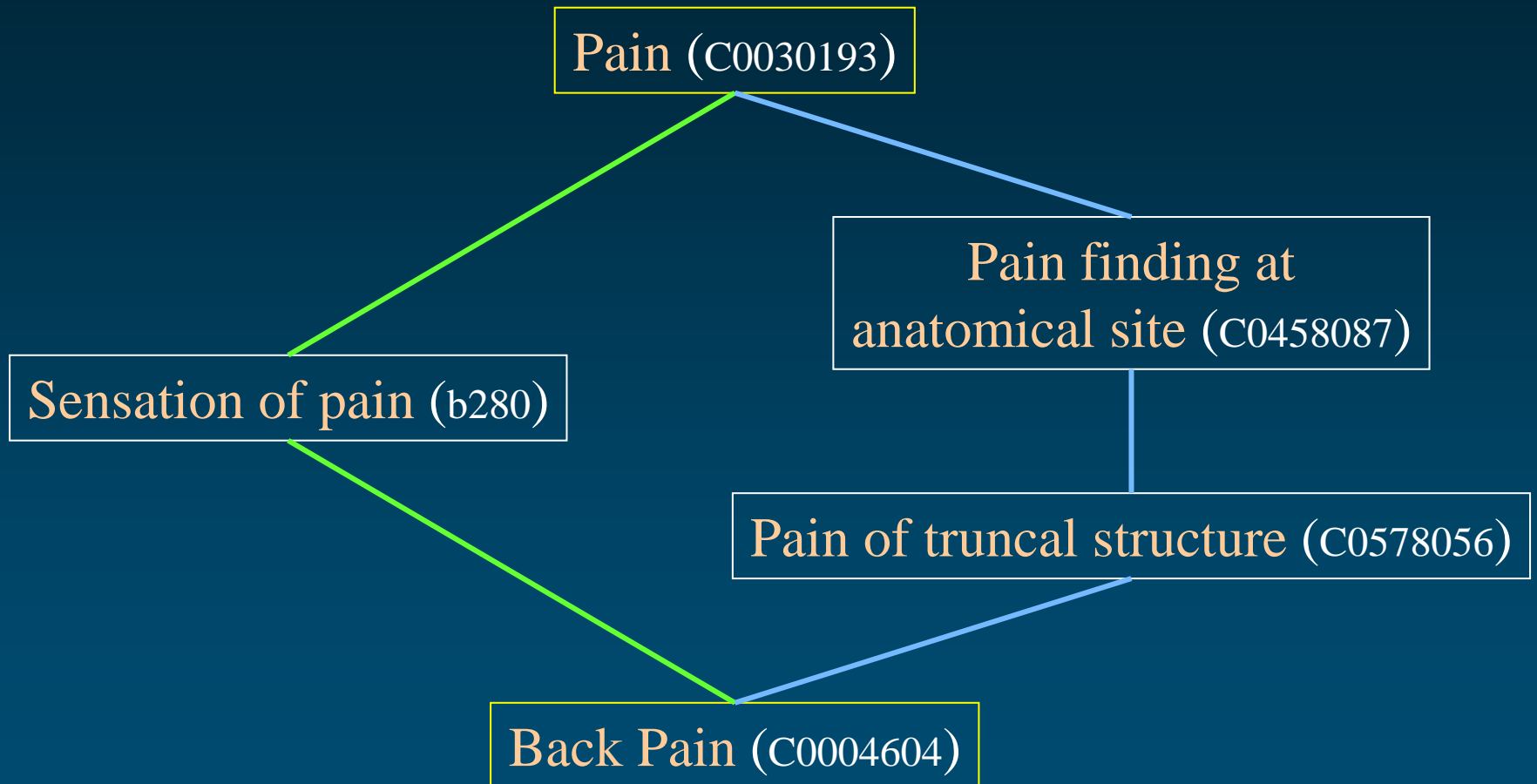


ICF relations in the Metathesaurus





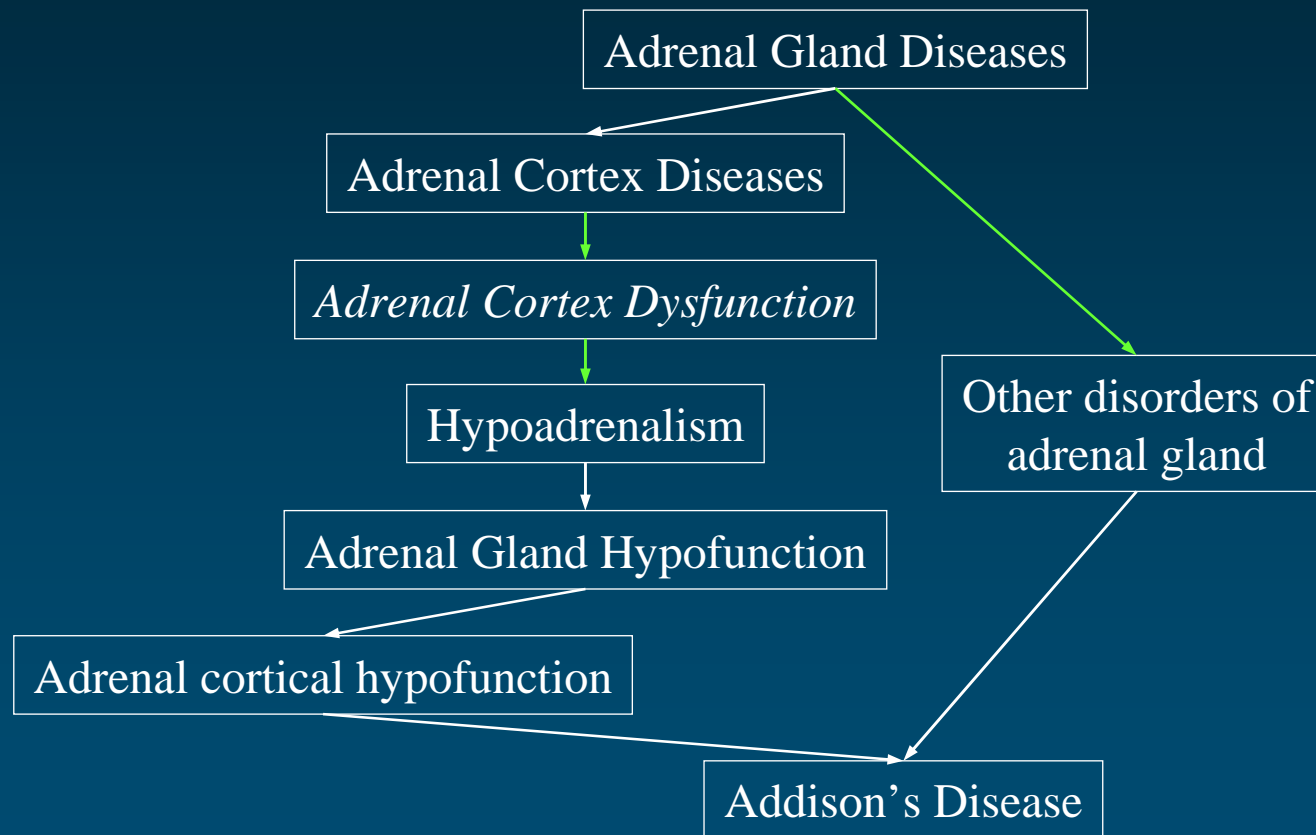
ICF relations in the Metathesaurus



Terminology integration methods

Editing and auditing

Additional knowledge: UMLS editors



Experience with ICF

Acknowledgments



- ◆ Marcy Harris
- ◆ Guergana Savova

Materials

◆ ICF: 1495 terms

- 478 terms filtered out

- 218 terms with *other specified*
- 217 terms with *unspecified*
- 37 terms with *other specified* and *unspecified*
- 2 terms with *specified* (alone)
- 1 term with *other specified* (alone)

- 1017 terms remaining

◆ UMLS: version 2004AA



Mapping to UMLS Metathesaurus

◆ Methods

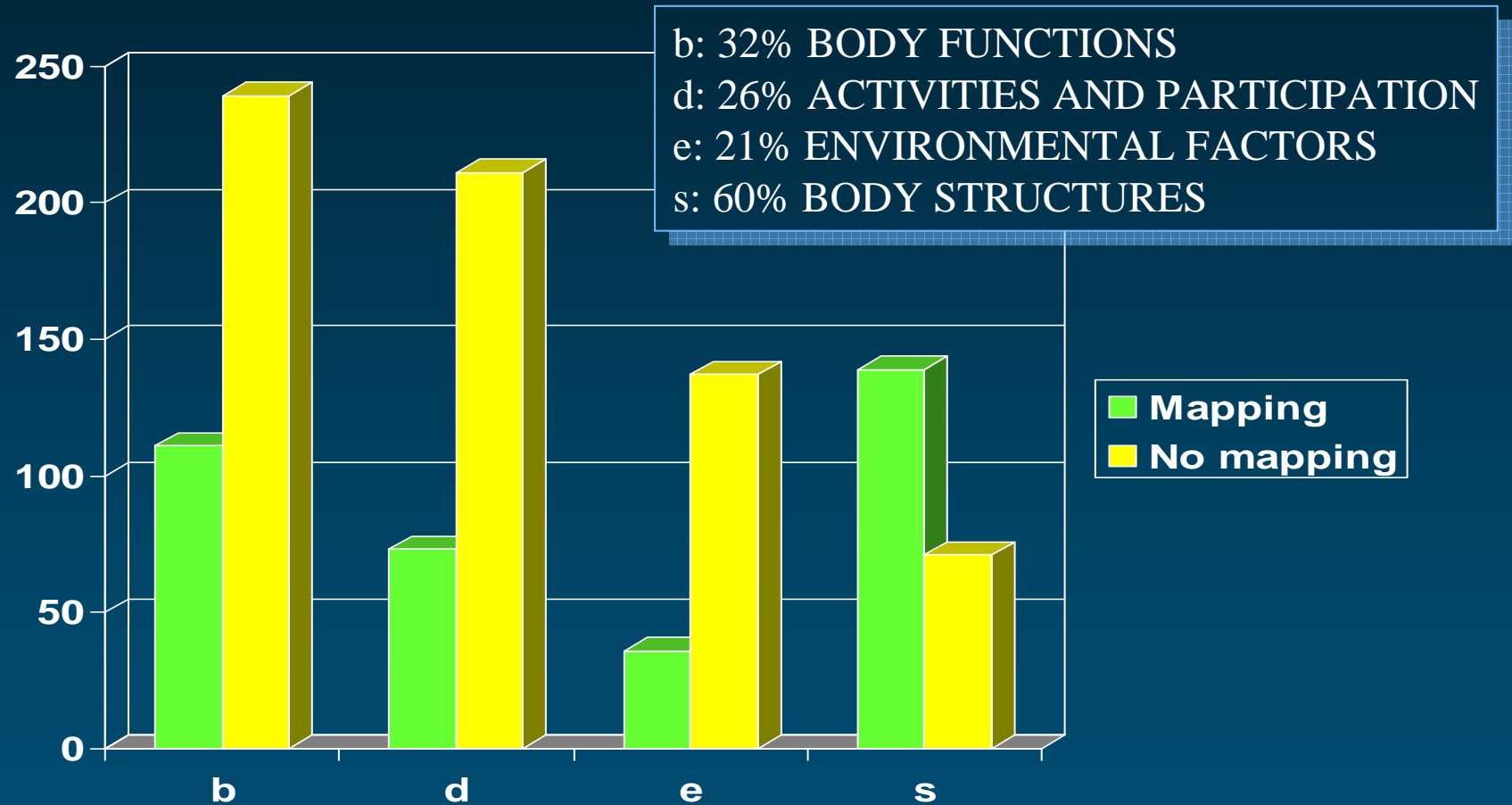
- Exact match first
- Normalized match, if necessary

◆ Results

- 359 ICF terms mapped (35%)
- 658 terms without mapping



Mapping by category



Issues with mapping

- ◆ Phenomena preventing the terms from being mapped:
 - coordination with *and* alone: 147
 - Education and training policies (e5852)
 - coordination with *or* alone: 7
 - Pain in stomach or abdomen (b28012)
 - coordination with both *and* and *or*: 2
 - Assistive products and technology for the practice of religion or spirituality (e1451)



Semantic validation

◆ Method

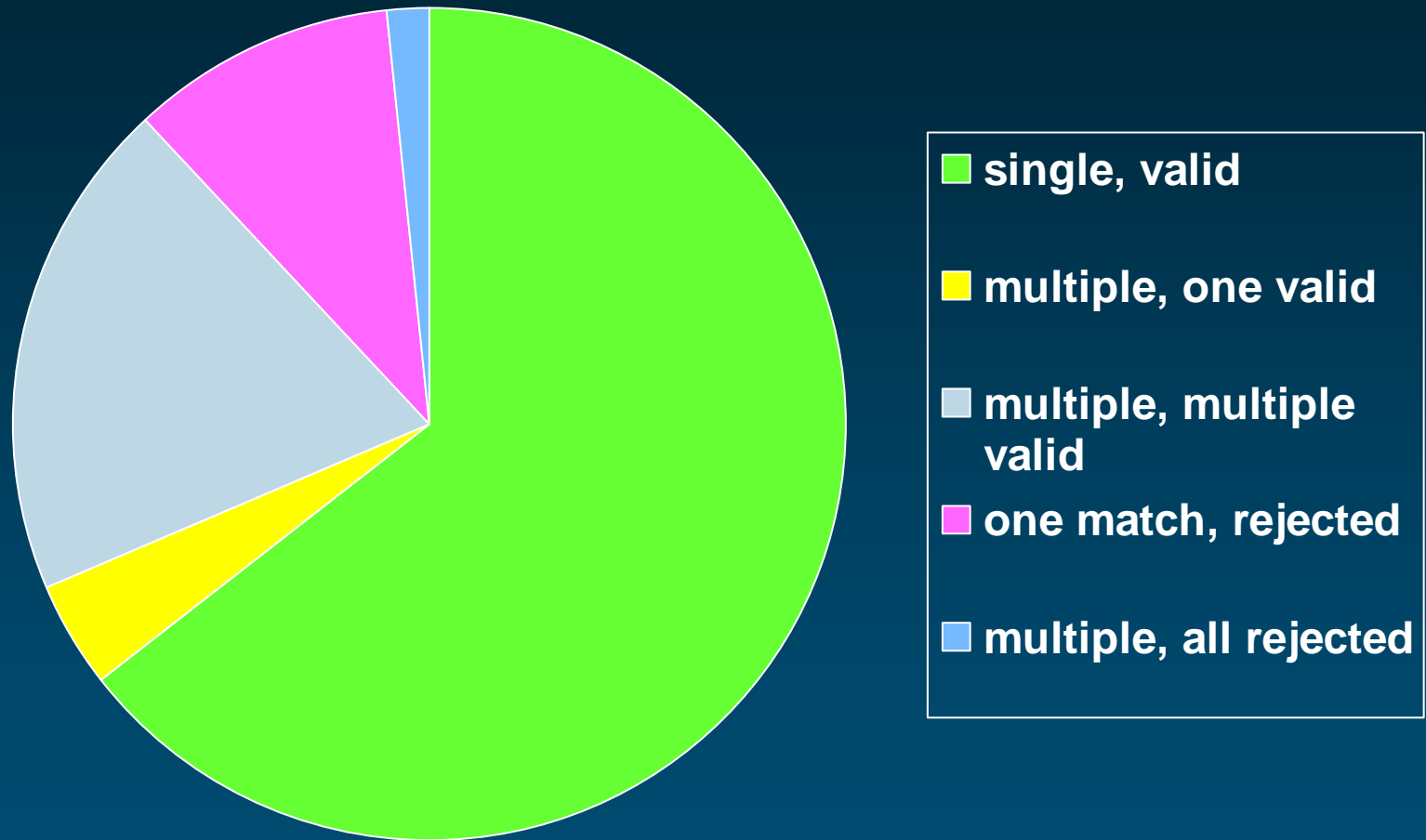
- Correspondence between ICF chapters and UMLS semantic types/groups

◆ Issues

- Correspondence difficult to establish for some subgroups in ENVIRONMENTAL FACTORS
 - PRODUCTS AND TECHNOLOGY
 - ATTITUDES



Semantic validation Results



Issues with semantic validation

- ◆ Multiple “valid” matches must be reviewed by experts and disambiguated
- ◆ Rejected mappings
 - Semantically invalid UMLS concepts
or
 - Missing correspondence (ICF chapter/UMLS ST-SG)



Conclusions

Conclusions (1)

- ◆ Integrating ICF into the UMLS
 - Should not be too difficult
 - Relatively small
 - Many concepts already present in UMLS
 - Challenges
 - Underspecified terms
 - Coordination
 - Specific perspective

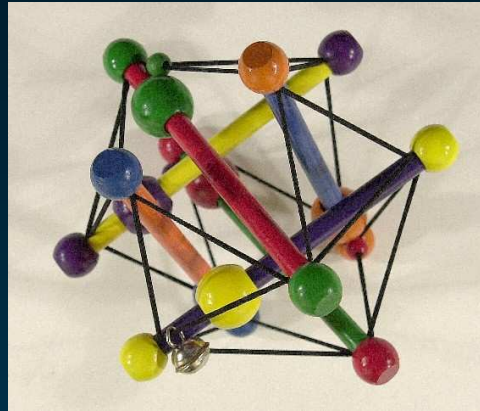


Conclusions (2)

◆ Integrating ICF into the UMLS

- Benefit for ICF
 - Links to other vocabularies
 - Facilitate downward extension
- Benefit for UMLS
 - Adds specific perspective





Medical Ontology Research

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